Bardet-Biedl Syndrome *GeneReview:* Molecular Genetics

Table 2. *BBS1* Pathologic Allelic Variants

Gene	Mutation	Exon	Reference
BBS1	p.E549X homozygote	16	Mykytyn et al 2002
BBS1	p.M390R homozygote	12	Mykytyn et al 2002
BBS1	p.M390R homozygote	12	Mykytyn et al 2002
BBS1	p.M390R heterozygote p.E549X heterozygote	12 16	Mykytyn et al 2002
BBS1	p.M390R heterozygote p.E549X heterozygote	12 16	Mykytyn et al 2002
BBS1	p.E549X heterozygote IVS4+1G>A	16 4	Mykytyn et al 2002
BBS1	p.Y284fsX288 homozygote	10	Mykytyn et al 2002
BBS1	p.M390R 27 homozygotes	12	Mykytyn et al 2002
BBS1	c.(-3)_37del heterozygote p.M390R heterozygote	1 12	Mykytyn et al 2003
BBS1	p.Y113X heterozygote p.M390R heterozygote	4 12	Mykytyn et al 2003
BBS1	V114fsX150 heterozygote p.L518P heterozygote	4 12	Mykytyn et al 2003
BBS1	p.I200_T201del p.M390R heterozygote	8 12	Mykytyn et al 2003
BBS1	p.Y284fsX288 heterozygote p.M390R heterozygote	16 12	Mykytyn et al 2003
BBS1	p.M347fsX373 heterozygote p.M390R heterozygote	11 12	Mykytyn et al 2003
BBS1	p.C377_F378delfsX412 homozygote	11	Mykytyn et al 2003
BBS1	p.R440X heterozygote p.M390R heterozygote	13 12	Mykytyn et al 2003
BBS1	p.L505fsX556 heterozygote p.M390R heterozygote	15 12	Mykytyn et al 2003
BBS1	p.L518P heterozygote p.M390R heterozygote	15 12	Mykytyn et al 2003
BBS1	p.H35R, 1 mutant allele		Beales et al 2003
BBS1	p.K53E, 1 mutant allele		Beales et al 2003
BBS1	p.L75fsX98, 1 mutant allele		Beales et al 2003

Gene	Mutation	Exon	Reference
BBS1	p.Y133X, 1 mutant allele		Beales et al 2003
BBS1	p.Q128X, 1 mutant allele		Beales et al 2003
BBS1	p.R146X, 4 mutant alleles		Beales et al 2003
BBS1	p.D148N, 4 mutant alleles		Beales et al 2003
BBS1	p.E234K, 1 mutant allele		Beales et al 2003
BBS1	IVS9-3C>G, 2 mutant alleles		Beales et al 2003
BBS1	p.Y284fsX288, 3 mutant alleles		Beales et al 2003
BBS1	p.Q291X, 1 mutant allele		Beales et al 2003
BBS1	p.G305S, 4 mutant alleles		Beales et al 2003
BBS1	p.389delI, 1 mutant allele		Beales et al 2003
BBS1	p.M390R, 74 mutant alleles		Beales et al 2003
BBS1	p.R429X, 1 mutant allele		Beales et al 2003
BBS1	p.Y434S, 1 mutant allele		Beales et al 2003
BBS1	p.R440X, 2 mutant alleles		Beales et al 2003
BBS1	IVS13-2A>G, 2 mutant alleles		Beales et al 2003
BBS1	p.R483X, 1 mutant allele		Beales et al 2003
BBS1	p.L503H, 1 mutant allele		Beales et al 2003
BBS1	p.L505fsX556, 1 mutant allele		Beales et al 2003
BBS1	p.L518Q, 1 mutant allele		Beales et al 2003
BBS1	p.L548fsX579, 1 mutant allele		Beales et al 2003
BBS1	p.E549X, 1 mutant allele		Beales et al 2003

.0001 BBS1, E549X. This mutation was found in homozygous form in all affected individuals from a consanguineous Puerto Rican family [Mykytyn et al 2002]. In addition, affected members of two other Puerto Rican families were compound heterozygotes with respect to E549X and M390R [Mykytyn et al 2002]. Futhermore, all affected individuals of a further Puerto Rican family were found to be compound heterozygous for E549X and a G to A transition at the +1 position of the splice donor site in exon 4 (IVS4+1G>A) [Mykytyn et al 2002]. This nonsense mutation was also identified in one mutant allele in a cohort of 259 individuals with BBS [Beales et al 2003].

.0002 BBS1, M390R. This mutation was identified in homozygous form in all affected members of a Puerto Rican family [Mykytyn et al 2002]. Two other

Puerto Rican families carried this mutation and the E549X in compound heterozygosity [Mykytyn et al 2002]. In addition, 22 out of 60 unrelated probands of mostly northern European ancestry with BBS had at least one copy; 16 were homozygous for the variant [Mykytyn et al 2002]. In a subsequent mutation survey, a total of 129 BBS probands were screened for the M390R mutation, 39 had at least one copy, 27 of whom were homozygous, indicating that this mutation was involved in 30% of the cohort [Mykytyn et al 2003]. In a further study of 259 individuals with BBS, a total of 74 M390R mutant alleles were identified, with M390R contributing to 18% of the cohort and involved in 79% of all families with BBS1 mutations [Beales et al 2003].

.0003 BBS1, IVS4+1G>A. All affected individuals of a Puerto Rican family were found to be compound heterozygotes for E549X and a G to A transition at the +1 position of the splice donor site in exon 4 [Mykytyn et al 2002].

.0004 BBS1, Y284fsX288. All affected members of a consanguineous Turkish family carried this frameshift mutation in homozygous form [Mykytyn et al 2002].

.0005 BBS1,c.(-3)_37del. This mutation was identified in compound heterozygous form with the M390R mutation in a BBS proband [Mykytyn et al 2003].

.0006 BBS1, p.Y113X. This nonsense mutation was identified in compound heterozygous form with the M390R mutation in a BBS proband [Mykytyn et al 2003].

.0007 BBS1, V114fsX150. This frameshift mutation was identified in compound heterozygous form with the L518P mutation in a BBS proband [Mykytyn et al. 2003].

.0008 BBS1,p.I200_T201del. This two amino acid deletion within exon 8 was identified in compound heterozygous form with the M390R mutation in an individual with BBS [Mykytyn et al 2003].

.0009 BBS1,p.Y284fsX288. This frameshift mutation was identified in compound heterozygous form with the M390R mutation in a BBS proband [Mykytyn et al 2003]. It was also identified in three mutant alleles in a cohort of 259 individuals with BBS [Beales et al 2003].

.0010 BBS1,p.M347fsX373. This frameshift mutation was identified in compound heterozygous form with the M390R mutation in a BBS proband [Mykytyn et al 2003]. It was also identified in three mutant alleles in a cohort of 259 individuals with BBS [Beales et al 2003].

.0011 BBS1,p.C377_F378delfsX412. This frameshift mutation was identified in homozygous form in a BBS proband [Mykytyn et al 2003].

.0012 BBS1, p.R440X heterozygote. This nonsense mutation was identified in compound heterozygous form with the M390R mutation in a BBS proband [Mykytyn et al 2003].

.0013 BBS1,p.L505fsX556. This frameshift mutation was identified in compound heterozygous form with the M390R mutation in a BBS proband [Mykytyn et al 2003].

.0014 BBS1, p.L518P. This amino acid substitution was identified in 3 BBS probands (Mykytyn et al. 2003). Two of these individuals were compound heterozygotes: one with the M390R mutation; the other with the V114fsX150 mutation [Mykytyn et al 2003].

.0015 BBS1, p.H35R. This mutation resulting in an amino acid substitution was identified in one mutant allele in a cohort of 259 individuals with BBS [Beales et al 2003].

.0016 BBS1, p.K53E. This mutation resulting in an amino acid substitution was identified in one mutant allele in a cohort of 259 individuals with BBS [Beales et al 2003].

.0017 BBS1, p.L75fsX98. This mutation resulting in a frameshift and the introduction of a premature stop codon was identified in one mutant allele in a cohort of 259 individuals with BBS [Beales et al 2003].

.0018 BBS1, p.Y113X. This nonsense mutation was identified in one mutant allele in a cohort of 259 individuals with BBS [Beales et al 2003].

.0019 BBS1, p.Q128X. This nonsense mutation was identified in one mutant allele in a cohort of 259 individuals with BBS [Beales et al 2003].

.0020 BBS1, p.R146X. This nonsense mutation was identified in four mutant alleles in a cohort of 259 individuals with BBS [Beales et al 2003].

.0021 BBS1, p.D148N. This mutation resulting in an amino acid substitution was identified in four mutant alleles in a cohort of 259 individuals with BBS [Beales et al 2003].

.0022 BBS1, p.E234K. This mutation resulting in an amino acid substitution was identified in one mutant allele in a cohort of 259 individuals with BBS [Beales et al 2003].

.0023 BBS1, IVS9-3C>G. A C to G nucleotide substitution was identified in the splice acceptor site of exon 10 in two mutant alleles in a cohort of 259 individuals with BBS [Beales et al 2003].

.0024 BBS1, p.Q291X. This nonsense mutation was identified in one mutant allele in a cohort of 259 individuals with BBS [Beales et al 2003].

.0025 BBS1, p.G305S. This mutation resulting in an amino acid substitution was identified in four mutant alleles in a cohort of 259 individuals with BBS [Beales et al 2003].

.0026 BBS1, p.389delI. This three base pair deletion resulting in the deletion of an isoleucine in exon 12 of BBS1 was identified in one mutant allele in a cohort of 259 individuals with BBS...0027 BBS1, p.R429X [Beales et al 2003].

.0028 BBS1, p.Y434S. This mutation resulting in an amino acid substitution was identified in four mutant alleles in a cohort of 259 individuals with BBS [Beales et al 2003].

.0029 BBS1, p.R440X. This nonsense mutation was identified in two mutant alleles in a cohort of 259 individuals with BBS [Beales et al 2003].

.0030 BBS1, IVS13-2A>G. This one base pair A to G substitution was identified in the splice acceptor site of exon 14 in two mutant alleles in a cohort of 259 individuals with BBS [Beales et al 2003].

.0031 BBS1, p.R483X. This nonsense mutation was identified in one mutant allele in a cohort of 259 individuals with BBS [Beales et al 2003].

.0032 BBS1, p.L503H. This mutation resulting in an amino acid substitution was identified in one mutant allele in a cohort of 259 individuals with BBS [Beales et al 2003].

.0033 BBS1, p.L505fsX556. This mutation resulting in a frameshift and the introduction of a premature stop codon was identified in one mutant allele in a cohort of 259 individuals with BBS [Beales et al 2003].

.0034 BBS1, pL518Q. This mutation resulting in an amino acid substitution was identified in one mutant allele in a cohort of 259 individuals with BBS [Beales et al 2003].

.0035 BBS1, p.L548fsX579. This mutation resulting in a frameshift and the introduction of a premature stop codon was identified in one mutant allele in a cohort of 259 individuals with BBS [Beales et al 2003].